



INDIANA UNIVERSITY

DEPARTMENT OF MEDICAL AND MOLECULAR GENETICS
School of Medicine

Announcement of Revised Diagnostic Testing

IU Genetic Testing Laboratories
Division of Diagnostic Genomics
April 5, 2018

This announcement pertains to testing for Huntington Disease testing in the IU Genetic Testing Laboratories. Detection of (CAG)_n expansion is used for both confirmatory testing of an individual exhibiting symptoms and for predictive testing in individuals who are at risk, but are not symptomatic.

Testing will not be performed for minors who are presymptomatic. Diagnostic testing may be completed with confirmation from the physician of symptoms present in a minor. All testing, regardless of age, requires an informed consent (attached).

It is widely recommended that predictive testing not be offered to individuals until they are at least 18 years old (or age of majority). The ethical principle of respect for autonomy dictates that individuals be allowed to make these decisions for themselves. Thus, an adult may decide to be tested. However, experience indicates that only about 10-15% of adult individuals at risk for HD choose to be tested (1). For that reason, and in the absence of a medical benefit for the child, it is important to protect the right of a child to decide for him or herself whether or not to pursue testing once they reach the age of majority. This position is supported by the American Society of Human Genetics, the American Academy of Pediatrics and the American College of Medical Genetics and Genomics (2-3).

References:

1. Sizer E, Haw T, Wessels TM et al (2012) The utilization and outcome of diagnostic, predictive and prenatal testing for Huntington disease in Johannesburg South Africa. *Genet Test and Mol Biomarkers* 16:58-62.
2. Botkin JR, Belmont JW, Berg JS, Berkman BE, Bombard Y, Holm IA, Levy HP, Ormond KE, Saal HM, Spinner NB, Wilfond BS and McInerney JD (2015) American Society of Human Genetics: Points to consider: ethical, legal and psychosocial implications of genetic testing in children and adolescents. *Am Journal of Hum Genet* 57: 1233.
3. Ross LF, Saal HM, David KL, Anderson, RR NS The American Academy of Pediatrics and the American College of Medical Genetics and Genomics (2013). *Genetics in Medicine* 15:234-235.

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